



Diamond-Blackfan Anemia

Answers to commonly asked questions



What is Diamond-Blackfan anemia?

Diamond-Blackfan anemia (DBA) is a rare blood disorder. In DBA, the bone marrow (the center of the bone where blood cells are made) does not make enough red blood cells that carry oxygen throughout the body. It is usually diagnosed during the first year of life.

What causes DBA?

Some people have a family history of the disorder. More than half of people with DBA have a known genetic cause. In many people with DBA, we do not know the cause.

How many new cases of DBA are diagnosed each year?

There are about 25 to 35 new cases of DBA each year in the United States and Canada.

Can anyone have DBA?

DBA affects both males and females equally. It occurs in every racial and ethnic group.

Can other members of the same family have DBA?

Yes, if someone has DBA there is up to a 50-percent chance that each of his or her children will have DBA. Symptoms can be very mild to very severe.

What are the signs and symptoms of DBA?

People with DBA have symptoms common to all other types of anemia, including pale skin, sleepiness, rapid heartbeat, and heart murmurs. In some cases, there are no obvious physical signs of DBA. However, about 30 to 47% of those with DBA have abnormal features involving the face, head, and hands (especially the thumbs). They might also have heart and kidney defects. Many children are short for their age and might start puberty later than normal.

Where did DBA get its name?

The anemia was named for Dr. Louis K. Diamond and Dr. Kenneth D. Blackfan, the first doctors who documented cases of the disease in the 1930s.



With good medical management, DBA patients can live long, healthy, active lives.

How do doctors know if someone has DBA?

Several tests can be used to tell if a person has DBA. One test a doctor can perform is called a bone marrow aspirate. This is a test in which a needle is inserted into a bone and a small amount of bone marrow fluid is taken out and studied under a microscope. Blood tests can also be done to see if there is a genetic basis for DBA or certain chemical abnormalities linked to DBA.

Will iron help reduce the anemia?

No; DBA is not caused by a lack of iron or other nutrients.

How is DBA treated?

To treat very low red blood cell counts in people with DBA, the two most common options are corticosteroids and blood transfusions. Bone marrow/stem cell transplantation might also be considered. Some people need no specific therapy. A qualified doctor can recommend the best treatment options.

What is corticosteroid treatment?

Corticosteroids are drugs used to treat many medical conditions. One type of corticosteroid is called oral prednisone, one of the most successful treatments for people with DBA.

What are the side effects of corticosteroid treatment?

Major side effects when these drugs are used in high doses for a long time include weight gain, water and salt retention, high blood pressure, muscle weakness, osteoporosis (brittle bones occasionally leading to fractures), wounds that won't heal, headaches, growth problems, eye diseases such as cataracts and glaucoma, a greater risk for infections, and the disruption of hormones that regulate normal body functions. Anyone using these drugs should be monitored carefully by a doctor.

What is a blood transfusion?

In a blood transfusion, a person receives healthy red blood cells from another person. With DBA, how often transfusions are needed varies from person to person. Transfusions can be needed as often as every 3 to 5 weeks, only a few times per year, or sometimes may not be needed for years.

Remember, you are not alone! There are hundreds of others around the world, just like you, living with DBA. We are here to help each other
- David's Mom

There are a number of DBA Specialists and support groups available; Scientific studies are ongoing and new treatment options are being made available.
- Julia

DIAMOND-BLACK FAN ANEMIA IS A TREATABLE, MANAGEABLE, AND SOMEDAY, A CURABLE DISORDER - Jose

Become your child's advocate, because many doctors are not familiar with DBA. Do your homework and become part of the decision-making process.
- Ashley's Dad



Living with DBA

People with DBA can live long, healthy, active lives if they get good medical care and live a healthy lifestyle. As long as their hemoglobin levels are high enough (hemoglobin is the substance in the red blood cells that supplies oxygen to all cells in the body), people with DBA can take part in all activities, usually without limitations.

Finding out that you or your child has DBA can be scary and overwhelming. Here are some tips to help you cope.

- **Contact the Diamond Blackfan Anemia Foundation.** To get in touch with families in the support network, see the back of this brochure.
- **Enroll in the Diamond Blackfan Anemia Registry.** This is a patient registry dedicated to collecting, analyzing, and sharing information on DBA. See the back of this brochure.
- **Join a support group.**
A list of online DBA support groups is included on the back of this brochure.
- **Learn as much as you can about DBA.**
If you don't understand certain medical terms and concepts, ask your doctor to explain. Take notes, tape conversations, and ask questions during doctor visits. Use a notebook to organize your medical information. Build a strong partnership with your medical team.
- **Stay positive.**
Research on DBA and other related conditions might soon lead to better treatments, new cures, and improved quality of life for people with DBA.
- **Talk with a mental health professional.**
Psychologists, social workers, and counselors can help you deal with the challenges of living with DBA or with someone in your family who has DBA.

Do blood transfusions have any complications?

Sometimes people can develop transfusion reactions, with symptoms such as fever and rash. Medication can be given before a transfusion to help prevent these symptoms. Red cell transfusions can also cause a buildup of extra iron in the body, which can harm the heart or liver, cause diabetes, slow down normal growth, and even cause death if the extra iron is not removed. The amount of iron in the body of a person with DBA must be checked regularly. If iron levels are too high, a doctor can recommend drugs to remove excess iron in body tissues. This process is called chelation therapy. People getting transfusions should avoid iron supplements.

What is bone marrow/stem cell transplantation?

Bone marrow/stem cell transplantation replaces a person's bone marrow/stem cells with those from a healthy, matching donor.

Are there other treatment options for DBA?

Other treatment options are being studied, but to date none work as well as corticosteroids or transfusion therapy. One day, a safe, reliable cure, perhaps using gene therapy, might be possible. However, this is still many years away.

Who can I call with questions?

For answers to your immediate medical questions, you can call the DBA nurse, Ellen Muir, RN, MSN, at 1-877-DBA-NURSE (322-6877) or the nurse at one of the DBA national resource centers near you. For additional information and support, call the DBA Foundation at 716-674-2818.



For information, networking, and support for yourself or someone else living with DBA, contact-

Foundations

Diamond Blackfan Anemia Foundation (DBAF)

Dedicated to providing support for DBA patients, families, and research. In partnership with DBA families, the DBAF funds worldwide research projects, provides educational materials and emotional support to families, networks DBA families, sponsors DBA family meetings including Camp Sunshine retreats (www.campsunshine.org), and publishes the comprehensive biannual DBA Newsletter and an informative monthly e-newsletter. DBAF is committed to educating, empowering, and supporting DBA patients and their families. For additional information and to register with the DBAF, please call the main number or visit www.dbafoundation.org.

P.O. Box 1092

West Seneca, New York 14224

Main Number: 716-674-2818

<http://www.dbafoundation.org>

<http://www.facebook.com/dbafoundation>

<http://www.twitter.com/dbafoundation>

Daniella Maria Arturi Foundation (DMAF)

Provides financial support for the research of DBA, increases awareness of DBA among medical professionals and federal policy makers, and coordinates and sponsors annual DBA scientific and clinical meetings. DMAF sponsors a comprehensive "DMA 4 DBA" annual newsletter as well as online news and e-blasts updating the DBA community about the latest in DBA news. Visit www.diamondblackfananemia.org to download current issues of the DMA 4 DBA newsletter and "subscribe" to be sure you receive all DBA news, events and updates.

P.O. Box 1434

Mattituck, NY 11952

<http://www.diamondblackfananemia.org/>

<http://www.facebook.com/daniellafoundation>

<http://www.twitter.com/dmafoundation>

Centers for Disease Control and Prevention (CDC)

Division of Blood Disorders

National Center on Birth Defects and

Developmental Disabilities

Centers for Disease Control and Prevention

1600 Clifton Road, MS E 64

Atlanta, GA 30333

Phone: 800-CDC-INFO (800-232-4636)

www.cdc.gov/dba

Registry

Diamond Blackfan Anemia Registry of North America (DBAR)

Established in 1993 to improve the understanding of DBA. The DBAR collects clinical data on DBA patients and their families and offers participation in research protocols.

Adrianna Vlachos, MD

Jeffrey M. Lipton, MD, PhD

Eva Atsidaftos, MA

Steven and Alexandra Cohen Children's Medical Center of New York

269-01 76th Ave., Room 255

New Hyde Park, NY 11040

Main number: 516-562-1504

Toll-free number: 1-888-884-DBAR (3227)

<http://www.dbar.org/>

DBA National Resource Centers

Steven and Alexandra Cohen Children's Medical Center of New York

Adrianna Vlachos, MD

Head, Bone Marrow Failure Program

Hematology/Oncology and Stem Cell

Transplantation

Director, Diamond Blackfan Anemia Registry

Surveillance and Awareness Program

Steven and Alexandra Cohen Children's Medical Center of New York

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Main number: 516-562-1598

Ellen Muir, RN, MSN

Clinical Nurse Specialist

Phone: 516-562-1505

DBA Nurse Hotline: 1-877-DBA-NURSE (322-6877)

University of Texas Southwestern Medical Center

Zora R. Rogers, MD

Professor and Clinical Director

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UT Southwestern Medical Center

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Web and E-mail-based Support Groups

Blackfan, an electronic family support group - Blackfan@yahoogroups.com

An e-mail server that allows you to send one e-mail that automatically gets distributed to everyone subscribed to the list. This is a semi moderated website and e-mail group for DBA parents and patients. To subscribe, e-mail: blackfansubscribe@yahoogroups.com, or go to the website at: <http://health.groups.yahoo.com/group/blackfan/>.

DBA21@yahoogroups.com

This is a semi moderated website and e-mail group for adult patients only. To subscribe, e-mail: dba21-subscribe@yahoogroups.com or go to the website at: <http://health.groups.yahoo.com/group/dba21/>.